December 2011

KANSAS NEWBORN SCREENING ANNUAL REPORT STATE FISCAL YEAR 2011 (JULY 2010—JUNE 2011)

Kansas Department of Health and Environment

Kansas Department of Health and Environment
1000 SW Jackson St.
Topeka, KS 66612

Special points of interest:

- New screening protocol for cystic fibrosis provides for significantly fewer false positives.
- Kansas identifies 73 infants with an inheritable disorder its newborn screening program.
- Quarterly newsletters implemented for better communication with stakeholders.
- NNSGRC provides financial review for NBS program.

Inside this issue:

GAVIN—A MOTHER'S STORY

Above: Newborn Gavin prior to newborn screening results

Happy and healthy, happy and healthy…that is what I prayed for every day of my pregnancy. After two miscarriages, my husband, Mark, and I were very excited for our little bundle of joy to arrive. Gavin James was born in Wichita, KS, on a warm November day in 2010. He weighed 6 pounds, 11 ounces and was 20 inches long. Gavin was tested for cystic fibrosis (CF) through the Kansas Newborn Screening Program when we were at the hospital, but that week was a "new baby blur" with little sleep, so we don't remember exactly when it took place. But such a quick and simple test would make a big difference for us in the weeks and months to come.

Above: Gavin at three months.

A few days after we arrived home from the hospital, we received a call from our pediatrician's office asking us to bring Gavin back in because one of his tests came back abnormal. We were scared and a bit sleep-deprived, but of course, we complied. CF didn't run in either of our families, so we were a bit confused. A few days (and many, many prayers) later, we received the call from Dr. Kinnane, our pediatrician, confirming the diagnosis of cystic fibrosis. We were shocked, devastated, scared and had so many questions. "What does this mean for Gavin? What do we do next?" We were quickly set up with an appointment to meet with Dr. Maria Riva at the CF Clinic in Wichita and to have Gavin undergo a sweat chloride test, which confirmed his diagnosis. After the first meeting with Dr. Riva, Gavin began a daily regimen of enzymes, inhalers, nebulizers and CPT (chest physical therapy). I couldn't believe my three week old baby was eating applesauce off of a spoon and taking enzymes with every meal – he wasn't even holding his head up yet!

As a hospital social worker and patient advocate by profession, I wanted to educate myself and my family about cystic fibrosis and find a way to get involved in the CF community. I volunteered for the Great Strides Walk as a committee member and was able to connect with other families affected by cystic fibrosis. Thanks to CF Social Worker, JoAnn, I was paired up with another CF mom who I could talk with and ask questions to. It was a tremendous help to speak with someone who really understood what I was going through as a new mom dealing with a new diagnosis.

At left: 11 month old Gavin with his mom, Karey.

I am so thankful for the Newborn Screen and the early detection of Gavin's cystic fibrosis. I believe he is growing and thriving today because preventative treatments were started prior to symptoms. We are also grateful for the incredible support from the CF Clinic staff and Mid-Kansas Pediatrics who have been on this journey with us since the beginning. Gavin just turned one year old, is crawling/pulling up with lightening speed and furniture walking, and is teething and drooling like crazy. He loves to eat, play with blocks and balls, has over 70 hats, and his smile lights up a room. And for now, he is exactly what we prayed for…happy and healthy!

See page 7 to learn more about the new cystic fibrosis screening test Kansas implemented in December 2010.

WHY NEWBORN SCREENING?

The goal of newborn screening is to identify and treat infants affected by a screened disorder so that disability, mental retardation and/or death can be prevented. All of the core metabolic disorders have treatments available. Although most of these disorders are rare, some occur as infrequently as 1:100,000, it remains important to screen, identify, and treat these infants early so that they can lead productive and healthy lives.

Newborn screening has been an integral part of an infant's health in Kansas since 1965 when testing for phenylketonuria (PKU) began. Since then the program has added additional tests, with the largest expansion beginning in July 2008 when Kansas added an additional twenty two disorders to their testing protocol. Kansas currently tests for 28 of the 29 metabolic disorders recommended by the American College of Medical Genetics (ACMG). Severe combined immune deficiency (SCID) was approved in May 2010. Ten states have implemented full or partial screening for SCID and another fifteen have approved SCID screening. Twenty-five states, including Kansas, are in a fact-finding stage regarding SCID screening. Jamey Kendall participates in the monthly SCID teleconference calls sponsored by ACMG and the Newborn Screening Translational Research Network (NBSTRN).

GOALS OF THE KANSAS NEWBORN SCREENING PROGRAM

Kansas has specific goals for its newborn screening program. These goals are:

- Ensure that each baby born in Kansas receives a newborn screening.
- Ensure that all infants with screened results that are outside of normal limits receive prompt and appropriate confirmatory testing.
- Ensure that all diagnosed individuals are referred for appropriate medical therapy.

How did we do in meeting our goals in SFY11?

- We tested 40,697 initial samples. Only one parent refused testing for their child based on religious reasons (the only reason a parent can refuse testing in Kansas). Because the newborn screening program is not currently linked with Kansas Vital Statistics, we are unable to ensure that every child born in Kansas was provided a newborn screening test. One goal for SFY11 was to have Kansas Health and Environmental Laboratories linked with the birth record to ensure that we test every child or a parent refusal form is obtained. This project will be piloted in SFY12.
- 100% of the primary care physicians (PCPs) for the 2780 infants who had results outside of normal limits were notified by both the newborn screening follow-up program coordinator and the neonatal testing laboratory. Of these, only 59 infants (2.1%) were lost to follow-up or did not follow up as recommended by the program.
- Seventy three infants were diagnosed with a metabolic disorder in SFY11. Twenty received services through the Children and Youth with Special Health Care Needs (CYSHCN) program at KDHE. Eleven attended a CYSHCN sponsored clinic and nine received direct services. All families with diagnosed infants are referred to CYSHCN, however not all families apply to this program, and not all families who apply are eligible to receive services through the program. Currently, Kansas does not have a formal long-term follow-up program to track infants identified with a genetic disorder through newborn screening. National stakeholders have identified newborn screening as an ideal case for the use of electronic health records. Kansas has challenges with its information reporting system as well as its capability to track long term outcomes for identified children.

PROGRAM OVERVIEW

The Kansas Newborn Screening (NBS) program is under the Kansas Department of Health and Environment (KDHE). The testing laboratory is located at Forbes Field in Topeka and the follow-up program is located at the Curtis State Office Building in downtown Topeka. The two sections of the program meet on a regular basis to discuss changes and to coordinate efforts within the program.

The laboratory is under the Health Chemistry section, lead by Stacey Sandstrom. Colleen Peterson is the manager of the laboratory and oversees the day-to-day operations. The laboratory technical staff includes June Carroll, Shawn Manos, Kathy Modin, Nestor Rodriguez, Laura Ross, and Christina Wiens. Customer service/Data Entry for the laboratory is done by Eugenia Akers, Katherine Appel, Rebecca Banka, Ron Peterson and Nancy Roberts (manager).

NBS follow-up is under the Bureau of Family Health, Children and Youth with Special Health Care Needs (CYSHCN) section at KDHE. CYSHCN is directed by Marc Shiff. The follow-up staff includes Jan Conklin and Diana Lopez, administrative assistants; Jamey Kendall and Linda Williams, follow-up coordinators; and Garry Kelley, epidemiologist.

Laboratory Staff, L to R: June Carroll, Colleen Peterson, Shawn Manos, Christine Wiens, Nestor Rodriguez, Kathy Modin, and Laura Ross.
Not pictured: Stacey Sandstrom.

Customer Service Staff, L to R: Katherine Appel, Eugenia Akers, Ron Peterson, Nancy Roberts.
Not pictured: Rebecca Banka.

Follow-up Staff, L to R: Jan Conklin, Linda Williams, Jamey Kendall, Garry Kelley and Diana Lopez.

ADVISORY COUNCIL

Kansas statute 65-180 states that the Secretary of KDHE shall appoint an advisory council to advise on the implementation of newborn screening. This council meets semi-annually in Topeka on the third Thursday of April and October. The meetings are open to the public. The FY2011 voting members were:

Lisa Butterfield, MS, CGC
Maternal Fetal Medicine, KU Medical Center

James Casey, MD
Pediatric Endocrinologist, Cotton-O'Neil Clinic

Dennis Cooley, MD
Pediatrician, Topeka

Diana Daldrup
March of Dimes

Majed Dasouki, MD—Council Chair
Pediatrics and Medical Genetics, KU Medical Center

Kenneth Dykstra, MD
Pediatric Endocrinologist, KUMC—Wichita

Brenda Issa, MD
Pediatrics, KUMC—Wichita

Michelle Leeker
Parent/Advocate

Catherine Fox, MS, RD, LD—Council Vice-Chair
KU Medical Center

Vance Lassey, MD
Family Medicine, Holton

Greta McFarland, MD
Pediatrician, Chanute

Glenn Edwards McGee, Ph.D.
Center for Practical Bioethics

Rebecca Reddy, MD
Pediatrician, KUMC—Wichita

William Randall Reed, MD
Neonatologist, Wesley Medical Center

Maria Riva, MD
Pediatric Pulmonologist, KUMC—Wichita

Mitzi Scotten, MD
KUMC Cystic Fibrosis Center

Margaret Smith, MD
KDHE—Health Finance

Deborah Stern, RN, JD
Kansas Hospital Association

Jakica Tancabelic, MD
Pediatric Hematologist, Cotton-O'Neil Clinic

Leona Therou, MD
Pediatrician, KUMC

Suzanne Wikle, MSW
Kansas Action for Children

A SYSTEMS APPROACH TO NEWBORN SCREENING

Newborn screening involves many partners to provide our newest citizens a quality newborn screen. Without these elements in place, the system would fail to provide the excellent service that Kansans have come to expect from newborn screening. These systems include:

- Education of healthcare providers and parents
- Collection of a quality sample
- Timely transport of specimen to the state laboratory
- Rapid and reliable testing methodology
- Timely notification of healthcare providers and parents of any unexpected result
- Timely repeat sample collection
- Appropriate referral of infant to specialists for diagnosis, treatment and counseling services
- Assuring access to programs that help our most vulnerable citizens
- Continuous quality improvement within the program

The Kansas Newborn Screening Program is committed to these systems and encourages our partners to develop, enhance and implement these systems as well.

NEWBORN SCREENING TEST RESULTS

Newborn Screening Follow-Up Results on Infants Screened in Kansas for SFY 2011

Condition Screened	Number of Presumptive Positive or Inconclusive Results on Initial Screen	Number of Normal Infants after repeat screen or other testing	Number of Pending Screen Results	Number of Screens Lost to Follow-Up to NBS	Number of Parental Notifications	Number of Deceased	Number of Confirmed Positive/ Diagnosed (classical or partial with treatment)
Biotinidase Deficiency	1	1	0	0	0	0	0
Cystic Fibrosis	279	233	0	0	6	2	9 Cystic Fibrosis 29 CF Carriers
Endocrine Disorders							
Congenital Adrenal Hyperplasia	87	79	0	1	0	3	4
Presumptive Congenital Hypothyroidism	62	40	0	1	0	0	21
Borderline Congenital Hypothyroidism	1001	939	0	46	0	0	15 + 1 transient CH
Galactosemia	5	4	0	0	0	0	1 Duarte
Hemoglobinopathies							
Sickle Cell Anemia	4	0	0	0	0	0	4
Sickle C Disease	4	0	0	0	0	0	4
Sickle/Beta-Thalassemia Disease	2	0	1	0	0	0	1
Other Hemoglobin Diseases	8	2 Hgb C Traits	1	0	1	0	1 Hgb E Disease 1 Sickle/α Thal 2 Hgb C/B Thal
Hemoglobin Traits	774	60	555	0	0	0	90 Sickle Traits 69 Other Traits
Amino Acid Disorders	330	312	1	2	1	11	1 ASA 1 PKU 1 MAT I/III[1]
Fatty Acid Oxidation Disorders	68	61	0	0	0	2	3 CUD 2 MCAD
Organic Acid Disorders	155	143	0	0	1	9	2 3MCC

1 = Methyladenosyltransferase deficiency; a secondary panel disorder

POSITIVE SCREENING RESULTS FOR SFY11

In SFY11, Kansas screened 40,697 infants. Of these 2,780 had presumptive positive or inconclusive results and required further testing. Most often, this meant the newborn screen had to be repeated. However, for certain presumptive positives, immediate consultation and additional blood work or urine analysis was indicated. For cystic fibrosis, a sweat chloride test was recommended if one or more mutations was detected.

On December 1, 2010 a new, two-tiered screening protocol was implemented for cystic fibrosis. Immune reactive trypsinogen (IRT) was still the initial screen used, however if the IRT was elevated, the same sample was refluxed to a polymerase chain reaction (PCR) test for cystic fibrosis transmembrane conductance regulator (CFTR) gene mutations. This reduced the number of infants needing a repeat NBS and also reduced the number of infants referred for a sweat chloride test. From 7/1/10 to 11/30/11 (five months of data) there were fifty nine infants referred for a sweat chloride. Of these, thirty four had normal sweat chloride (normal infants), twelve were identified as CF carriers and two were diagnosed with cystic fibrosis. After implementation of IRT/DNA, (12/1/10 to 6/30/11—six months of data) only twenty nine infants were referred for a sweat chloride test. Of these, three who had high IRTs (≥170 ng/mL) but no mutations detected were normal infants, seventeen were identified as CF carriers and six were diagnosed with cystic fibrosis. Three infants (with a single mutation) are pending and parents have been notified of the need to complete a sweat chloride test.

One thousand sixty three infants had positive results for congenital hypothyroidism (CH) - 62 were presumptive positive and 1001 were borderline. Thirty seven were diagnosed, including one with transient CH. Of the 37 diagnosed, 15 of those had initial borderline results (including the one transient diagnosis).

Seven hundred ninety two hemoglobin results were reported to physicians for follow-up. Of these eighteen were presumptive hemoglobin diseases and seven hundred seventy four were hemoglobin traits. Approximately 20% of the traits have been confirmed, while another five hundred fifty five are still pending. The recommendation is to do confirmatory testing at the one year exam, so this pending number is not unexpected.

There were five hundred fifty three abnormal MS/MS results—three hundred thirty with amino acid disorders, sixty eight with fatty acid oxidation disorders and one hundred fifty five with organic acid disorders. Ninety three per cent were confirmed as normal infants after either a repeat screen or additional testing. Three infants were diagnosed with an amino acid disorder—one with argininosuccinic aciduria (ASA), one with phenylketonuria (PKU) and one with Methyladenosyltransferase deficiency (MAT). MAT is on the secondary screening panel but can be identified by an elevated methionine level, which is also the marker for homocystinuria (HCY) a core panel disorder. Five fatty acid oxidation disorders were diagnosed—three carnitine uptake defect (CUD) and two medium chain acyl-coA dehydrogenase deficiency (MCAD). Two infants were diagnosed with 3-methylcrotonyl CoA carboxylase deficiency (3MCC), an organic acid disorder.

NEWBORN SCREENING TESTS PERFORMED IN KANSAS

MISC. DISORDERS
BIO - Biotinidase Deficiency
CAH - Congenital Adrenal Hyperplasia
CH - Congenital Hypothyroidism
CF - Cystic Fibrosis
GALT - Galactosemia

AMINO ACID DISORDERS
ASA - Argininosuccinic Acidemia
CIT - Citrullinemia
HCY - Homocystinuria
MSUD - Maple Syrup Urine Disease
PKU - Phenylketonuria
TYR - Tyrosinemia Type I

ORGANIC ACID DISORDERS
IVA - Isovaleric Acidemia
GA-1 - Gluteric Acidemia Type I
HMG - 3-hydroxy-3-methylglutaryl-CoA lyase deficiency
MCD - Multiple Carboxylase Deficiency
MMA (MUT) - Methylmalonic Acidemia - Mutase Deficiency
3MCC - 3-Methylcrotonyl-CoA Carboxylase Deficiency
MMA (CBL A,B) - Methylmalonic acidemia - cblA & cblB
PROP - Proprionic Acidemia
BKT - Beta-ketothiolase Deficiency

HEMOGLOBINOPATHIES
SCA - Sickle Cell Anemia
HB S/TH - HB-S/Beta Thalassemia
HB S/C - HB-S/C Disease

FATTY ACID DISORDERS
CUD - Carnitine Uptake Defect
LCHAD - Long Chain 3-OH Acyl-CoA-Dehydrogenase Deficiency
MCAD - Medium Chain Acyl-CoA Dehydrogenase Deficiency
VLCAD - Very Long Chain Acyl-CoA Dehydrogenase Deficiency
TFP - Tri Functional Protein Deficiency

NEWBORN SCREENING INITIATIVES IN FY11

Above: Linda Williams and Laura Ross at the training in Lawrence.

Education

- Five regional trainings for collection facilities were held in June 2011. Training locations were Hays, Garden City, Overland Park, Lawrence and Wichita. One hundred eight professional staff members from over forty -seven facilities associated with newborn screening attended the trainings. The focus was to engage attendees as partners, and to provide them with the necessary tools to educate parents and healthcare providers about newborn screening. Linda Williams and Laura Ross were the instructors for the trainings.

Staff Development

- Jamey Kendall and Colleen Peterson attended a SCID conference sponsored by CDC in Atlanta, GA in October 2010.
- Jamey Kendall attended the Effective Long-term Follow-up (LTFU) conference in Washington D.C. sponsored by American College of Medical Genetics (ACMG) in March 2011.
- Linda Williams was sponsored by CDC to attend the National Birth Defects Prevention Network's 14th annual meeting in Orlando, FL in February 2011.
- Laura Ross attended the MS/MS Laboratory Workshop, hosted by the Association of Public Health Laboratories (APHL) in Raleigh, NC in May 2011.
- Christine Wiens attended the Newborn Screening Molecular Training Workshop, sponsored by APHL and CDC in Atlanta, GA in June 2011.
- June Carroll and Shawn Manos each attended a Region 4 training at Mayo Clinic in Rochester, MN.
- Colleen Peterson attended the Evaluation of IRT as a Biomarker for Cystic Fibrosis meeting, sponsored by National Newborn Screening and Genetics Resource Center (NNSGRC), APHL, CDC, and Health Resources and Services Administration (HRSA) in Annapolis, MD in May 2011.
- Jamey Kendall and June Carroll attended a one day workshop on Lysosomal Storage Disorders at Children's Mercy Hospitals and Clinics.

Laboratory Improvements

- To decrease the overall turn-around time for samples received on Friday, the laboratory now has a technician work Sunday evening to set up the biotinidase deficiency assay and begin the sixteen hour incubation time. This allows the test to be read on Monday and reduced turn-around times for initial results from three or more working days to only one working day!
- The laboratory has contracted with ChemWare to implement a new Lab Information Management System (LIMS) for the entire lab.

Heartland Genetics and Newborn Screening Collaborative

- The KS NBS program continues to be an effective participant with the Heartland Genetics and Newborn Screening Collaborative (HGNBSC). Staff members participate in monthly Newborn Screening Workgroup calls. Jamey Kendall is a member of the HGNBSC Advisory Council. Linda Williams was a member of the HBNBSC Transition Workgroup.
- Four program staff attended the annual HGNBSC meeting in Des Moines, IA in September 2010. The meeting was an opportunity to network with newborn screening and genetics professionals within the Heartland states.
- Four program staff attended the HCNBSC Newborn Screening Workgroup meeting in Kansas City, MO in April 2011.
- HGNBSC funded a parent support liaison (PSL) project at two Children and Youth with Special Health Care Needs clinics. This was in coordination with the Systems in Sync program within the CYSHCN program.

NEW SCREENING PROTOCOL FOR CYSTIC FIBROSIS

On December 1st, 2010, the Kansas Health and Environmental Laboratories (KHEL) went live with a new screening protocol for cystic fibrosis (CF) using blood spot cards. KHEL performed a three month pilot project prior to implementation. The previous protocol used for CF was IRT/IRT in which the immune-reactive trypsinogen (IRT) levels were tested on an initial blood spot card. If the specimen IRT levels were elevated, a second specimen was requested. A sweat chloride test was recommended if the second specimen also had abnormal levels of IRT to determine if the infant had cystic fibrosis.

The new protocol is known as IRT/DNA. KHEL determines the IRT levels using only the initial blood spot card. If the confirmed IRT levels are elevated above normal; a DNA test is performed. The DNA test looks for mutations within the cystic fibrosis trans-membrane regulator (CFTR) gene. The panel tested at KHEL includes 40 of the most common CF mutations, including the 23 mutations recommended by the American College of Medical Genetics (see chart below). A sweat chloride test is performed at an accredited Cystic Fibrosis Center when one or more mutations are detected. In Kansas, CF Centers are located at KU Medical Center in Kansas City and Via Christi in Wichita. A sweat chloride test is also recommended if the IRT level is ≥ 170 ng/mL without any detected DNA mutations.

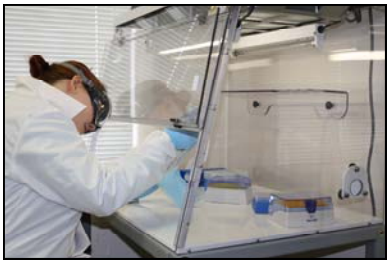
When using the IRT/IRT method for cystic fibrosis screening, over 90% of the cases that were called abnormal ended up being false positives. During FY 2009 161 repeat screens had elevated IRT values. Of these only 10 were diagnosed with cystic fibrosis and 4 were found to be CF carriers. Similarly in FY 2010 there were 114 repeat screens with an elevated IRT value; 10 of these were confirmed as having cystic fibrosis and no carriers were noted.

Using the IRT/IRT method caused much unnecessary worry for many parents of newborns, added to the number of repeat screens being requested, sent many infants for sweat tests, and also created some bad press for the Newborn Screening Program.

From December 1, 2010 to June 30th, 2011 there were 233 specimens with an elevated IRT (≥60 ng/mL) that were tested with the new IRT/DNA method. Of these specimens 207 had no mutations detected. There were 21 that had one heterozygous mutation (usually are carriers), two that had two heterozygous mutations (possibly indicative of CF), one equivocal result, and two that showed homozygous mutations (usually indicative for CF). Seventeen were confirmed as carriers (including the equivocal result), and six were confirmed as diagnostic CF cases.

The new protocol has:

- Reduced the number of required repeat blood spot samples
- Reduced the number of infants sent for a sweat chloride test
- Decreased the turn-around time for the infant sweat test referral
- Reduced the time to a diagnosis
- Reduced the undue concern for many parents of newborns
- Reduced the number of false positives



Above: Laura Ross, NBS Lab staff member pipettes newborn screening samples for DNA amplification..

CFTR InPlexTM assay 40-mutation panel

▲F508	R1162X	2184delA	394delTT	D1152H
G542X	**3120+1G>A**	**3659delC**	E60X	1078delT
W1282X	**R117H**	**A455E**	Q493X	S549R T>G
G551D	**1717-1G>A**	**R560T**	3905insT	Y1092X C>G
621+1G>T	**2789+5G>A**	**G85E**	V520F	Y1092X C>A
N1303K	**R347P**	**1898+1G>A**	S549R A>C	2183AA>G
R553X	**711+1G>T**	**3849+10kbC>T**	Y122X	S549N
▲I507	**R334W**	3876delA	R347H	3849+4A>G

(ACMG recommended mutations in **bold**)

Blood Spot Quality

The quality of the blood spot card (BSC) samples submitted to the laboratory for testing continues to be one of the top priorities for process improvement. The FY10 annual unsatisfactory blood spot percent was 4.17. In FY11 the unsatisfactory rate dropped to 3.07 percent through continued training and communication with our collection facilities. It is useful to determine which facilities continue to make improvements in this crucial area. When analyzing this data, it is helpful to compare similar sized facilities. The unsatisfactory goal of less than 2.0 percent has not been met, therefore, continued efforts are being made in this area. Below are facilities that had a 25 percent or greater decrease in the number of unsatisfactory blood spot samples submitted for testing in FY11 compared to FY10.

FACILITIES WITH > 999 SAMPLES IN SFY11

Facility Name	Facility ID	SFY10 BSC Unsat rate	SFY11 BSC unsat rate	SFY11 Total samples	SFY11 # of BSC unsats	Change
SAINT FRANCIS HOSPITAL	1760	1.5	0.6	1030	6	-62.1
MENORAH MEDICAL PARK	6060	6.6	2.6	1117	29	-60.6
LAWRENCE MEMORIAL HOSPITAL	430	2.7	1.1	1139	13	-57.3
MERCY REGIONAL HEALTH CENTER	1570	2.9	1.5	1102	17	-46.4
STORMONT-VAIL REG MED CTR	1770	3.5	2.3	2323	54	-32.8
KANSAS UNIV MED CTR	2060	1.3	0.9	1902	17	-31.3
WESLEY MEDICAL CTR	1660	3.8	2.8	6874	191	-27.1
SAINT JOSEPH MEDICAL CTR	1670	5.2	3.8	2603	100	-26.2

FACILITIES WITH 500 - 999 SAMPLES IN SFY11

Facility Name	Facility ID	SFY10 BSC unsat rate	SFY11 BSC unsat rate	SFY11 Total samples	SFY11 # of BSC unsats	Change
SOUTHWEST MEDICAL CENTER	1740	4.7	2.1	679	14	-55.8
HAYS MEDICAL CENTER	6660	1.3	0.8	732	6	-37.3
VIA CHRISTI HOSP PITTSBURG INC	380	4.7	3.0	539	16	-36.8
NEWTON MEDICAL CENTER	740	3.2	2.3	572	13	-28.8

FACILITIES WITH 100 - 499 SAMPLES IN SFY11

Facility Name	Facility ID	SFY10 BSC unsat rate	SFY11 BSC unsat rate	SFY11 Total samples	SFY11 # of BSC unsats	Change
WILLIAM NEWTON MEM HOSP	360	2.8	0.4	280	1	-87.4
COFFEYVILLE REG MED CTR	1240	9.7	1.9	214	4	-80.7
NEOSHO MEMORIAL HOSPITAL	1330	4.4	0.9	338	3	-79.6
MEMORIAL HOSPITAL-MCPHERSON	1140	5.2	1.1	174	2	-78.0
MITCHELL COUNTY HOSPITAL HS	1200	21.2	5.2	116	6	-75.6
PEDIATRICS ASSN OF OLATHE	6500	7.1	2.0	396	8	-71.5
BIRTH & WOMENS CENTER	1820	19.6	5.9	170	10	-70.0
SO CENTRAL KS REG MED CENTER	350	10.9	4.0	149	6	-63.2
SUSAN B ALLEN MEM HOSP	170	2.4	1.0	311	3	-59.0
LABETTE HEALTH	950	7.1	3.0	232	7	-57.4
MERCY & TRUTH MED MISSIONS-KC	10505	28.2	15.0	100	15	-46.8
GREAT BEND REGIONAL HOSPITAL	92	7.1	4.1	270	11	-42.7
ATCHISON HOSPITAL	40	6.2	3.7	191	7	-40.7
CITIZENS MEDICAL CTR	1950	6.0	3.8	132	5	-36.7
NEWMAN REGIONAL HEALTH	1070	5.4	3.7	410	15	-32.5
SAINT JOHNS HOSPITAL	970	9.5	6.8	117	8	-28.2

FACILITIES WITH 50 - 99 SAMPLES IN SFY11

Facility Name	Facility ID	SFY10 BSC unsat rate	SFY11 BSC unsat rate	SFY11 Total samples	SFY11 # of BSC unsats	Change
HIAWATHA COMM HOSP	140	4.2	0.0	99	0	-100.0
ONAGA COMMUNITY HOSPITAL	1460	5.4	0.0	76	0	-100.0
KATHY BRACE MIDWIFE	8000	7.3	2.0	50	1	-72.5
SABETHA COMMUNITY HOSPITAL,INC	1300	4.8	2.0	50	1	-58.0
CLAY CO MEDICAL CENTER	280	12.5	8.5	71	6	-32.4
NORTON CO HOSPITAL	1370	22.4	16.2	68	11	-27.7

FACILITIES WITH 10 - 49 SAMPLES IN FY11

Facility Name	Facility ID	SFY10 BSC unsat rate	SFY11 BSC unsat rate	SFY11 Total samples	SFY11 # of BSC unsats	Change
MORRIS CO HOSPITAL	1260	2.0	0.0	40	0	-100.0
BIRTH & WOMENS HEALTH CENTER	8203	5.9	0.0	37	0	-100.0
MID-KS PEDIATRIC ASSOC P.A.	6400	7.1	0.0	29	0	-100.0
WICHITA CLINIC-NORTHEAST	7090	17.0	0.0	20	0	-100.0
WICHITA CLINIC - CARRIAGE PKWY	5060	4.5	0.0	13	0	-100.0
HOSPITAL DIST #1 OF RICE CO	1540	16.4	2.4	41	1	-85.1
STANTON CO HOSPITAL	1890	14.3	3.4	29	1	-75.9
ROOKS COUNTY HEALTH CENTER	1580	18.9	5.3	19	1	-72.2
MERCY HOSPITAL-MOUNDRIDGE	1120	10.3	2.9	34	1	-71.3
OLATHE FAMILY PHYSICIANS	10506	25.0	10.0	10	1	-60.0
HEIDGEN AND MILLS, PA	5950	15.4	7.7	13	1	-50.0
WASHINGTON CO HOSPITAL	1980	27.3	14.3	14	2	-47.6
CLOUD CO HEALTH CTR	290	5.6	3.0	33	1	-45.5
SMITH CO MEMORIAL HOSPITAL	1860	10.0	5.9	34	2	-41.2
PEDIATRIC CARE SPECIALISTS,PA	6550	10.3	6.7	30	2	-35.0
CHILDREN'S MERCY WEST	5570	6.7	4.3	23	1	-34.8

TOP TEN FACILITIES (>100 SAMPLES) WITH HIGHEST % IMPROVEMENT RATE FROM FY10 to FY11

Facility Name	Facility ID	SFY10 BSC Unsat rate	SFY11 BSC unsat rate	SFY11 Total samples	SFY11 # of BSC unsats	Change
WILLIAM NEWTON MEM HOSP	360	2.8	0.4	280	1	-87.4
COFFEYVILLE REG MED CTR	1240	9.7	1.9	214	4	-80.7
NEOSHO MEMORIAL HOSPITAL	1330	4.4	0.9	338	3	-79.6
MEMORIAL HOSPITAL-MCPHERSON	1140	5.2	1.1	174	2	-78.0
MITCHELL COUNTY HOSPITAL HS	1200	21.2	5.2	116	6	-75.6
PEDIATRICS ASSN OF OLATHE	6500	7.1	2.0	396	8	-71.5
BIRTH & WOMENS CENTER	1820	19.6	5.9	170	10	-70.0
SO CENTRAL KS REG MED CENTER	350	10.9	4.0	149	6	-63.2
SAINT FRANCIS HOSPITAL	1760	1.5	0.6	1030	6	-62.1
MENORAH MEDICAL PARK	6060	6.6	2.6	1117	29	-60.6

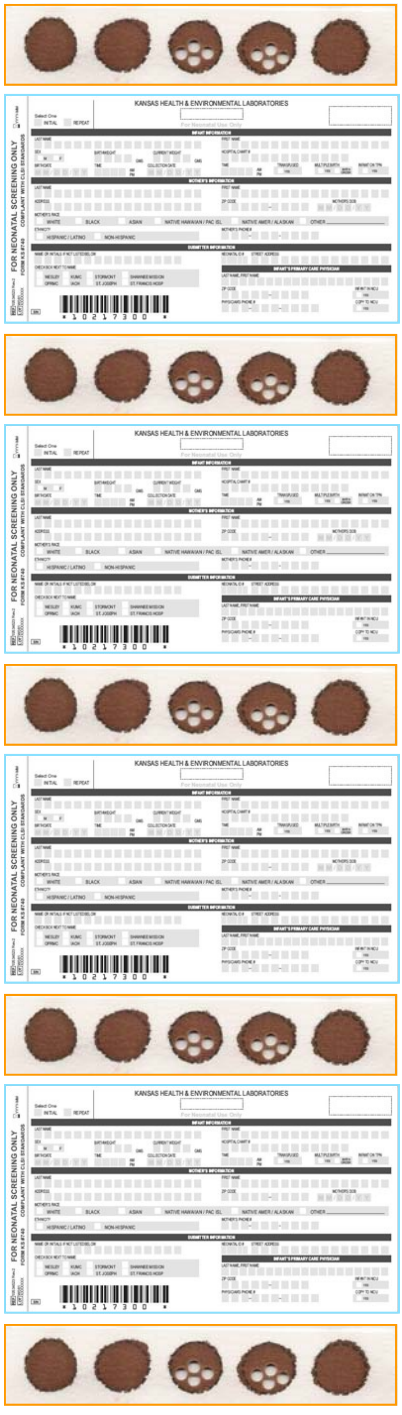
Eighty of the 152 (52.6%) submitting facilities had a 25% or greater reduction in their blood spot errors. However, 46 facilities (30.3%) had an increase in the number of unsatisfactory blood spots when compared to FY10. This latter group includes one of the larger birthing facilities (1380 births) in the state and 41 facilities that had less than 10 samples submitted in FY11.

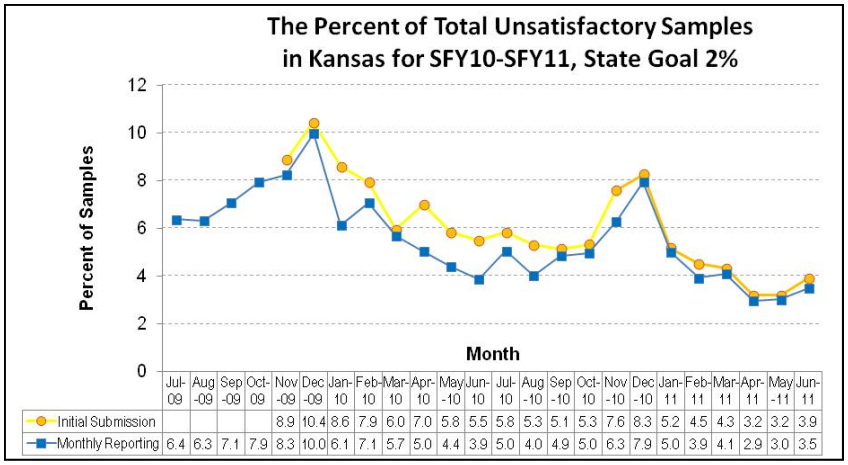
Of the 44,907 samples submitted for testing (includes repeat samples), 1,378 samples were unsatisfactory due to the quality of the blood spots. In addition, 923 samples had missing demographic information and 899 were drawn at <24 hours. NICU samples accounted for 40.4% of the <24 hour samples.

Monthly Unsatisfactory Samples

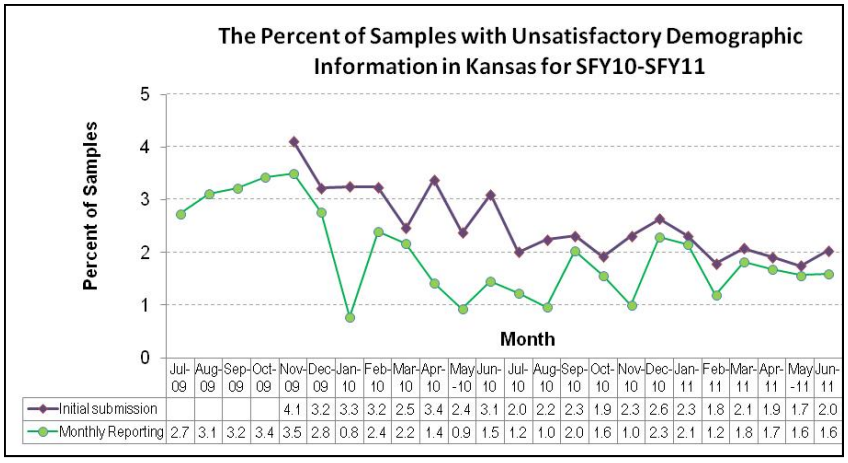
Since July 2009 the total percentage of unsatisfactory samples has continued to decline. In July 2009 the overall unsatisfactory rate was 6.4 percent. In June 2011, the overall unsatisfactory rate was 3.5 percent. Blood spot errors continue to decrease. In July 2009, the blood spot error rate was 3.6 percent and in June 2011 the rate was 1.9 percent. The previous goal of an overall unsatisfactory rate of below2.0 percent remains unattained. For FY12, a new goal of 1.3 percent has been established. This is based on the 2010 national average of 1.3 percent. The NBS staff continue to educate and communicate with collection facilities to support meeting this goal.

Follow-up staff continue to track demographic unsatisfactory samples received by the lab. Monthly collection facility reports are completed two weeks after the close of the month. By this time, many of the demographic errors have been resolved so the initial unsatisfactory rate is not reflected in the report. The graphs below show the monthly unsatisfactory rates, including the initial and end of month (reported) rates.

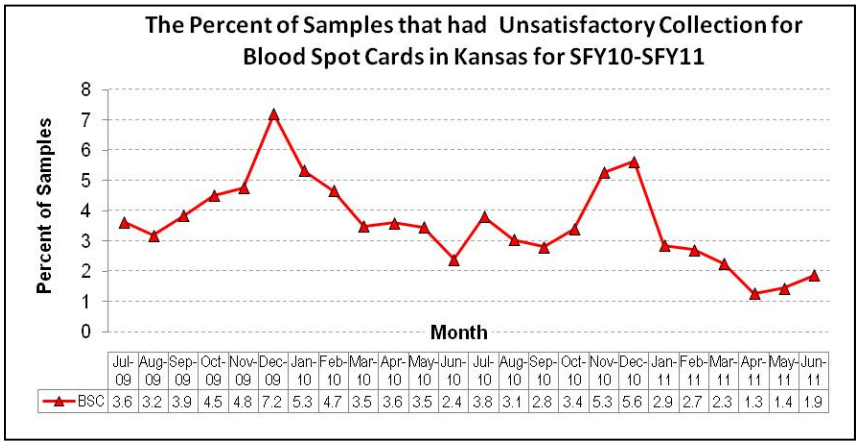




	Jul-09	Aug-09	Sep-09	Oct-09	Nov-09	Dec-09	Jan-10	Feb-10	Mar-10	Apr-10	May-10	Jun-10	Jul-10	Aug-10	Sep-10	Oct-10	Nov-10	Dec-10	Jan-11	Feb-11	Mar-11	Apr-11	May-11	Jun-11
Initial Submission					8.9	10.4	8.6	7.9	6.0	7.0	5.8	5.5	5.8	5.3	5.1	5.3	7.6	8.3	5.2	4.5	4.3	3.2	3.2	3.9
Monthly Reporting	6.4	6.3	7.1	7.9	8.3	10.0	6.1	7.1	5.7	5.0	4.4	3.9	5.0	4.0	4.9	5.0	6.3	7.9	5.0	3.9	4.1	2.9	3.0	3.5



	Jul-09	Aug-09	Sep-09	Oct-09	Nov-09	Dec-09	Jan-10	Feb-10	Mar-10	Apr-10	May-10	Jun-10	Jul-10	Aug-10	Sep-10	Oct-10	Nov-10	Dec-10	Jan-11	Feb-11	Mar-11	Apr-11	May-11	Jun-11
Initial submission					4.1	3.2	3.3	3.2	2.5	3.4	2.4	3.1	2.0	2.2	2.3	1.9	2.3	2.6	2.3	1.8	2.1	1.9	1.7	2.0
Monthly Reporting	2.7	3.1	3.2	3.4	3.5	2.8	0.8	2.4	2.2	1.4	0.9	1.5	1.2	1.0	2.0	1.6	1.0	2.3	2.1	1.2	1.8	1.7	1.6	1.6

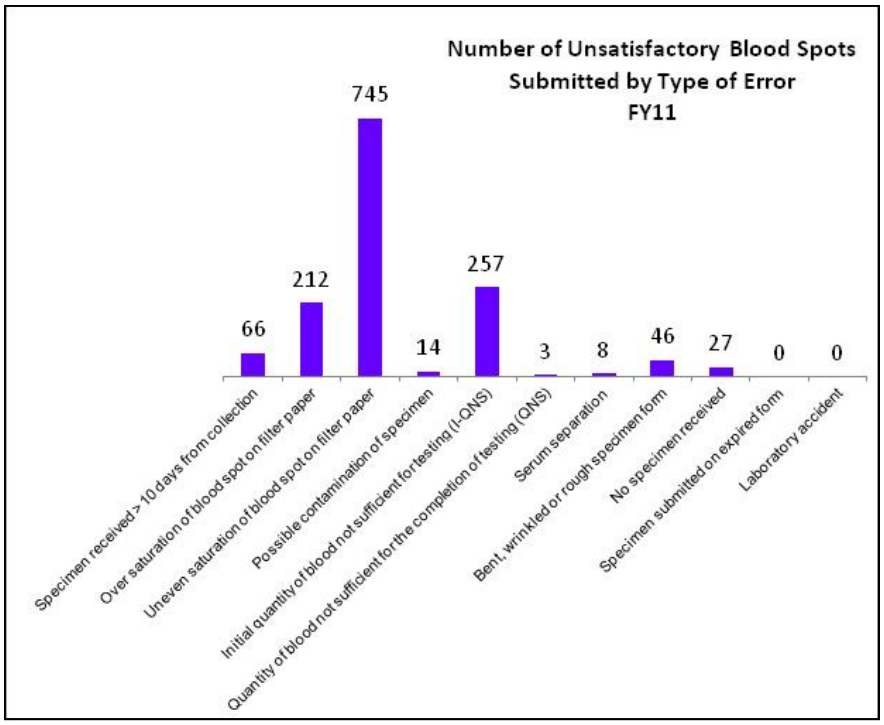


	Jul-09	Aug-09	Sep-09	Oct-09	Nov-09	Dec-09	Jan-10	Feb-10	Mar-10	Apr-10	May-10	Jun-10	Jul-10	Aug-10	Sep-10	Oct-10	Nov-10	Dec-10	Jan-11	Feb-11	Mar-11	Apr-11	May-11	Jun-11
BSC	3.6	3.2	3.9	4.5	4.8	7.2	5.3	4.7	3.5	3.6	3.5	2.4	3.8	3.1	2.8	3.4	5.3	5.6	2.9	2.7	2.3	1.3	1.4	1.9

Reasons For Unsatisfactory Blood Spot Samples

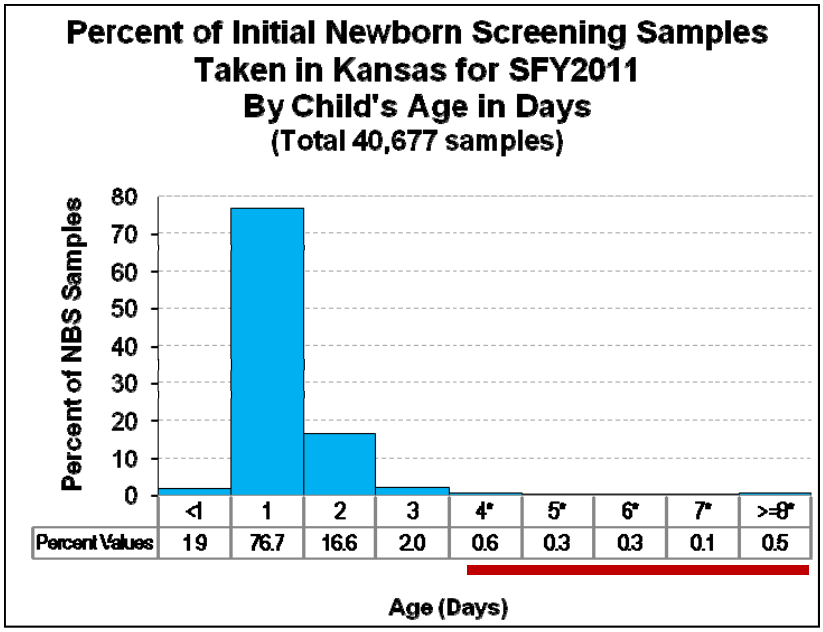
The most common reason for an unsatisfactory blood spot is the uneven saturation of the blood sample. The laboratory currently uses thirty reason codes when rejecting a sample; those codes can be grouped into eleven common reasons.

1) Specimen > 10 days old
2) Oversaturation of blood spot
3) Uneven saturation of blood spot
4) Possible contamination of specimen
5) Initial quantity not sufficient for testing (QNS)
6) Quantity not sufficient for completion of testing
7) Serum separation
8) Bent, wrinkled or rough specimen form
9) No specimen submitted
10) Specimen submitted on expired form
11) Laboratory accident



Age of Infant at Time of Initial Sample Collection

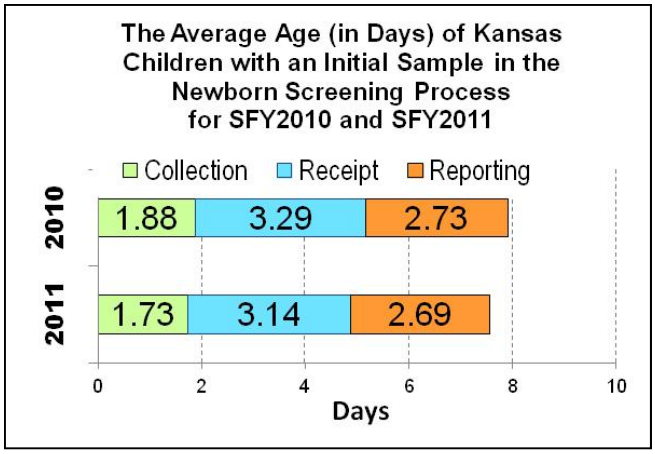
The Kansas Newborn Screening Advisory Council recommends that the initial sample for newborn screening be collected between 24 and 72 hours after birth. In SFY2011, 97.2 percent of initial samples were collected within this time frame.



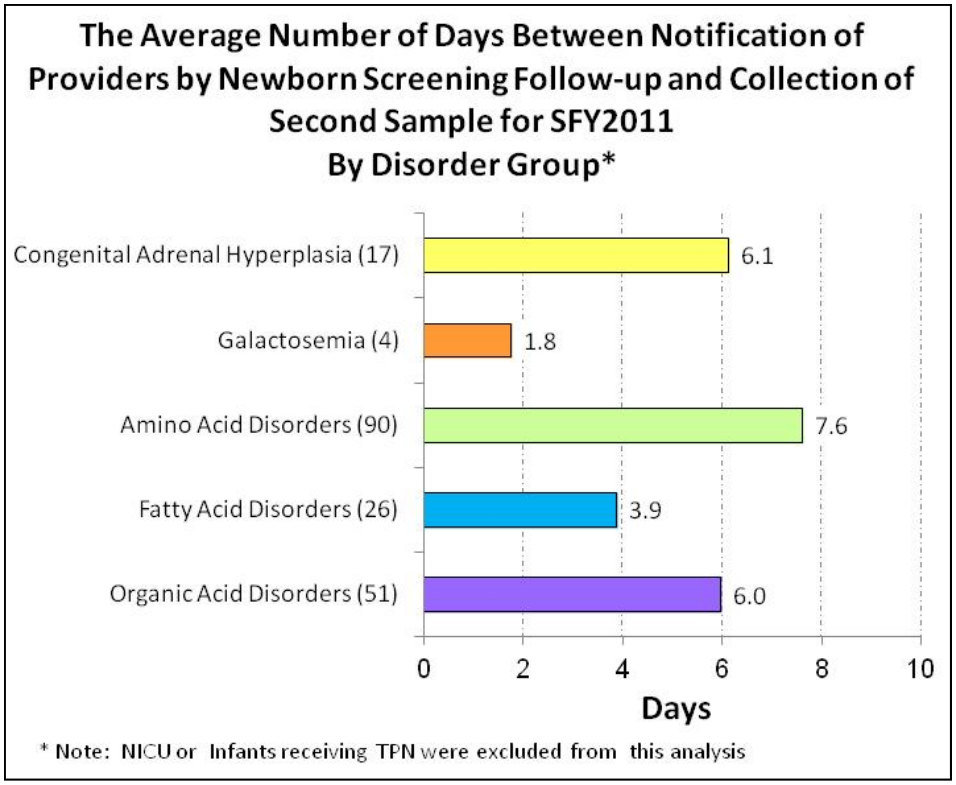
Turn Around Times

Time can be critical in newborn screening. It's important to: (1) track the time it takes to collect a sample at the birthing facility, (2) the length of time it takes to get that sample to the laboratory, and (3) how long it takes the lab to get abnormal results to the follow-up team.

In FY11, the average age at sample collection was 1.73 days. It took an average of 3.14 days from date of collection to date of receipt at the laboratory. When the lab took receipt of the sample, it took an average of 2.69 days to report an abnormal result to the follow-up team. This is an improvement from SFY2010.

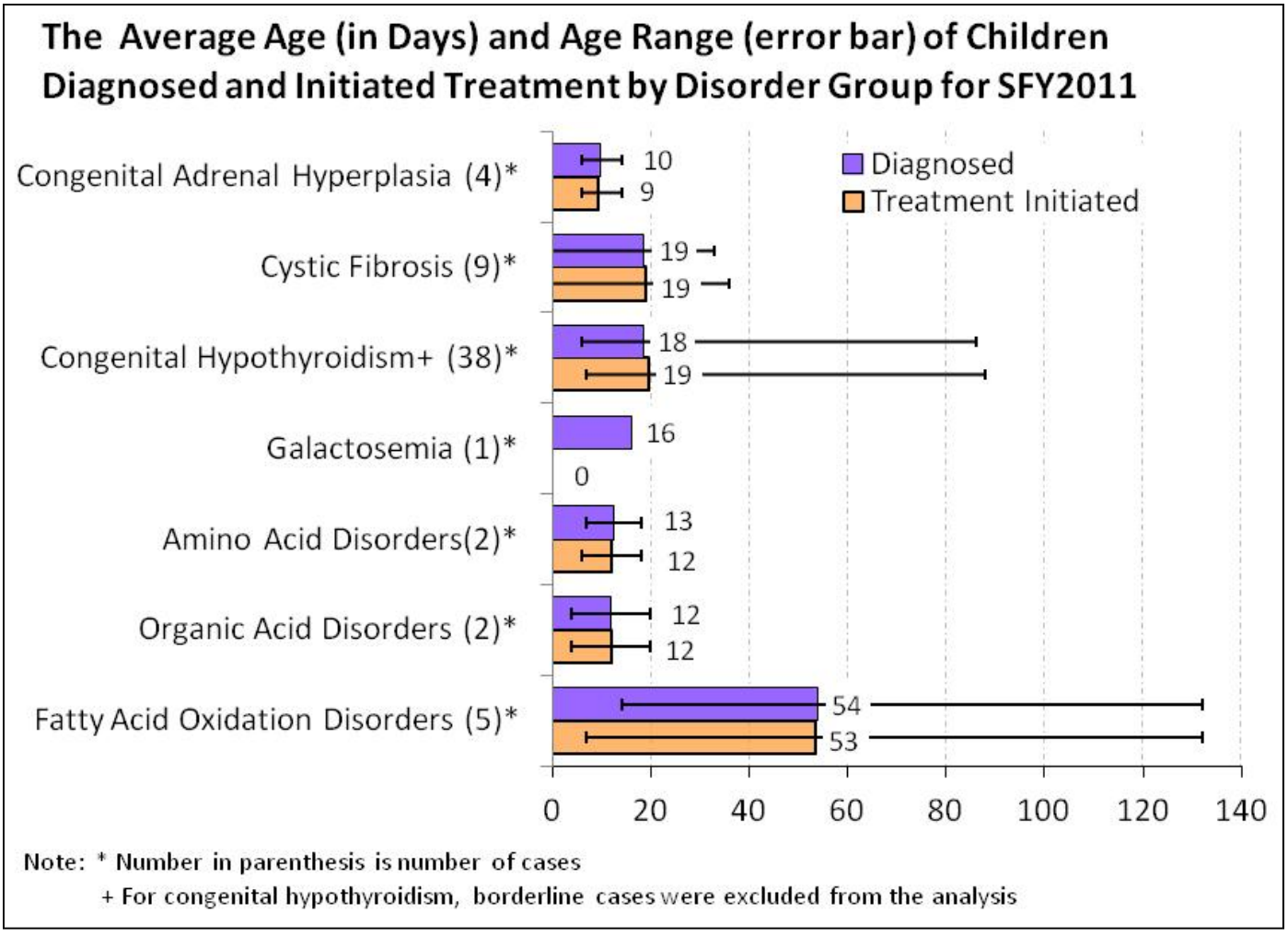


A second important timeframe is the length of time it takes the provider to get a requested repeat sample collected for retest when the initial result is abnormal. In the table below, NICU and infants known to be receiving TPN were excluded because many NICU infants have an automatic repeat sample collected, whether or not a repeat sample was requested. We wanted to capture how long it takes primary care physicians to contact the parent and arrange to have a second sample collected. Why some conditions seem to take longer to recollect is unclear.



Diagnosis and Treatment

When an infant is identified with an abnormal newborn screen, typically the screen is repeated prior to recommending diagnostic testing. Some presumptive positives go directly to diagnostic testing. The goal of the program is to quickly identify those infants who need treatment for a diagnosed condition. Treatment often begins prior to the completion of all diagnostic testing, as seen in the table below; especially if a diagnostic test needs to be sent to a reference lab. As seen in the table below, for most conditions the average time to diagnosis and treatment is less than three weeks of age. The one exception is fatty acid disorders. One infant was not screened until one month of age, which added to the average age at diagnosis.



COMMUNICATION

In FY2011 the Newborn Screening Program recognized the need for consistent and effective communication with our stakeholders. To that end, most collection facilities were contacted by phone to locate a staff member in both the nursery and laboratory as the point of contact for that facility. Phone numbers and email addresses were collected. This has improved our ability to electronically contact a large group of stakeholders.


KANSAS NEWBORN SCREENING QUARTERLY NEWSLETTER

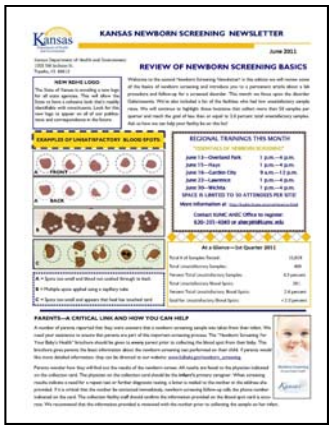
Welcome to the 1st Newborn Screening Newsletter!

DID YOU KNOW?

EXAMPLE OF GOOD BLOOD SPOTS:

New Screening Protocol for Cystic Fibrosis Implemented on December 1, 2010

In March 2011 the first Kansas Newborn Screening Quarterly Newsletter was published and sent electronically to collection facilities as well as members of the Kansas Chapter of American Academy of Pediatrics (KAAP) and Kansas Academy of Family Physicians (KAFP). The response has been extremely positive. The newsletters have provided the program with an easy mechanism to convey important information. Each newsletter highlights those facilities who have been successful in meeting the state goal of <2.0 percent unsatisfactory samples. It also includes an article on a lab procedure put into layman's terms. Newsletters are archived and available on our website at www.kdheks.gov/newborn_screening.


KANSAS NEWBORN SCREENING NEWSLETTER

REVIEW OF NEWBORN SCREENING BASICS

NEW NBS LOGO

EXAMPLES OF UNSATISFACTORY BLOOD SPOTS:

REGIONAL TRAININGS THIS MONTH

PARENTS—A CRITICAL LINK AND HOW YOU CAN HELP

To improve communication within the NBS program, a staff exchange occurred between KHEL NBS laboratory staff members and KDHE NBS follow-up staff members. Follow-up staff each spent a full day at the NBS laboratory at Forbes Field, observing the work flow to better understand the process from receipt of a sample to reporting of results. Similarly, KHEL NBS laboratory staff members each spent a day at the Curtis State Office Building with NBS follow-up staff to become familiar with how follow-up is done for repeat sample requests due to abnormal results or an unsatisfactory blood spot specimen, how additional testing is documented and how all final diagnosis for abnormal results are tracked.

NEWBORN SCREENING ANNUAL BUDGET AND EXPENDITURES

In FY11 the Kansas Newborn Screening Program received funding from two sources: 1) Children's Initiative Fund (CIF) and 2) Maternal Child Health Block Grant (MCH). The laboratory and follow-up programs have separate budgets within the program. The newborn screening laboratory received funding only from CIF and not MCH. Follow-up received funding from both CIF and MCH. Each year the Children's Cabinet, who administers CIF funds conducts an evaluation of the program through the University of Kansas to ensure that the program is effective and a good steward of the funds received through CIF. The table below indicates the funding source, amounts and whether it was allocated to the laboratory or follow-up program.

FY11 NEWBORN SCREENING BUDGET AND EXPENDITURES									
FUNDING SOURCE	Budgeted			Expended			Balance		
	Laboratory	Follow-up	Total	Laboratory	Follow-up	Total	Laboratory	Follow-up	Total
Children's Initiative Funds	$ 1,897,345.00	$ 321,098.00	$ 2,218,443.00	$ 1,885,170.94	$ 292,672.39	$ 2,177,843.33	$ 12,174.06	$ 28,425.61	$ 40,599.67
Maternal Child Health Block Grant	$ -	$ 115,765.00	$ 115,765.00	$ -	$ 93,495.68	$ 93,495.68	$ -	$ 22,269.32	$ 22,269.32
Total of Sources	$ 1,897,345.00	$ 436,863.00	$ 2,334,208.00	$ 1,885,170.94	$ 386,168.07	$ 2,271,339.01	$ 12,174.06	$ 50,694.93	$ 62,868.99

EXTERNAL PROGRAM REVIEW

On December 13-14, 2010, an external Newborn Screening Financial Review Team met with Kansas NBS staff members to review program finances and make related recommendations. Members of the review team were Dr. Brad Therrell, Jr. from the National Newborn Screening and Genetics Resource Center (NNSGRC), Dr. Harry Hannon, NNSGRC, Julie Luedtke, Nebraska Newborn Screening and Genetics Program and Dr. Gary Hoffman, Wisconsin Newborn Screening Laboratory. This review was made possible through a cooperative agreement with the Genetic Services Branch of the Maternal and Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA).

Intensive interviews occurred with Kansas NBS staff members regarding all aspects of the current program including laboratory operations, information technology, education, follow-up operations, assistance to families for diagnosis and treatment and financial resources available to the program.

The review team praised the Kansas program and the dedication of its staff to "providing a fiscally sound, quality newborn screening program that meets the needs of Kansas citizens". The review team also recommended several areas for improvement:

- Consideration of a fee for testing.
- Need for a comprehensive data management system, to include a statutorily required disease registry.
- Need for improved transport of specimens to state laboratory, including consideration of a courier system.
- Consideration of extended laboratory hours to include a weekend and holiday shift.
- Need for a dedicated education specialist.
- Consideration of adding severe combined immune deficiency (SCID) to KS panel, which was added to the ACMG core panel in May 2010.

A copy of the complete report (Dec 2010 KS Fiscal Review) is available at www.kdheks.gov/newborn_screening/links_interest.html

UPCOMING INITIATIVES/GOALS FOR SFY12

Education/Communication

- Education and outreach will be done with family practitioners so parents are informed about newborn screening prior to the birth of their child.
- Continued education for collection facilities to lower the number of unsatisfactory blood spot card submissions.

Information Systems

- Integrate laboratory information system (Informix) with Vital Statistics so the NBS program can track every child born in Kansas.
- Begin implementation of Laboratory Information Managements System (LIMS) for Kansas Health and Environmental Laboratories (KHEL).

Laboratory

- Implement a new reporting protocol for samples collected before 24 hours of age. This should reduce the number of false positive results for CAH, CH and IRT.